achondroplasia

Achondroplasia is a form of short-limbed dwarfism. The word achondroplasia literally means "without cartilage formation." Cartilage is a tough but flexible tissue that makes up much of the skeleton during early development. However, in achondroplasia the problem is not in forming cartilage but in converting it to bone (a process called ossification), particularly in the long bones of the arms and legs. Achondroplasia is similar to another skeletal disorder called hypochondroplasia, but the features of achondroplasia tend to be more severe.

All people with achondroplasia have short stature. The average height of an adult male with achondroplasia is 131 centimeters (4 feet, 4 inches), and the average height for adult females is 124 centimeters (4 feet, 1 inch). Characteristic features of achondroplasia include an average-size trunk, short arms and legs with particularly short upper arms and thighs, limited range of motion at the elbows, and an enlarged head (macrocephaly) with a prominent forehead. Fingers are typically short and the ring finger and middle finger may diverge, giving the hand a three-pronged (trident) appearance. People with achondroplasia are generally of normal intelligence.

Health problems commonly associated with achondroplasia include episodes in which breathing slows or stops for short periods (apnea), obesity, and recurrent ear infections. In childhood, individuals with the condition usually develop a pronounced and permanent sway of the lower back (lordosis) and bowed legs. Some affected people also develop abnormal front-to-back curvature of the spine (kyphosis) and back pain. A potentially serious complication of achondroplasia is spinal stenosis, which is a narrowing of the spinal canal that can pinch (compress) the upper part of the spinal cord. Spinal stenosis is associated with pain, tingling, and weakness in the legs that can cause difficulty with walking. Another uncommon but serious complication of achondroplasia is hydrocephalus, which is a buildup of fluid in the brain in affected children that can lead to increased head size and related brain abnormalities.

Frequency

Achondroplasia is the most common type of short-limbed dwarfism. The condition occurs in 1 in 15,000 to 40,000 newborns.

Genetic Changes

Mutations in the *FGFR3* gene cause achondroplasia. The *FGFR3* gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. Two specific mutations in the *FGFR3* gene are responsible for almost all cases of achondroplasia. Researchers believe that these mutations cause the

FGFR3 protein to be overly active, which interferes with skeletal development and leads to the disturbances in bone growth seen with this disorder.

Inheritance Pattern

Achondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. About 80 percent of people with achondroplasia have average-size parents; these cases result from new mutations in the *FGFR3* gene. In the remaining cases, people with achondroplasia have inherited an altered *FGFR3* gene from one or two affected parents. Individuals who inherit two altered copies of this gene typically have a severe form of achondroplasia that causes extreme shortening of the bones and an underdeveloped rib cage. These individuals are usually stillborn or die shortly after birth from respiratory failure.

Other Names for This Condition

- ACH
- achondroplastic dwarfism
- dwarf, achondroplastic

Diagnosis & Management

These resources address the diagnosis or management of achondroplasia:

- GeneReview: Achondroplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1152
- Genetic Testing Registry: Achondroplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001080/
- MedlinePlus Encyclopedia: Achondroplasia
  https://medlineplus.gov/ency/article/001577.htm
- MedlinePlus Encyclopedia: Hydrocephalus
  https://medlineplus.gov/ency/article/001571.htm
- MedlinePlus Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm
- MedlinePlus Encyclopedia: Spinal Stenosis
  https://medlineplus.gov/ency/article/000441.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Achondroplasia
  https://medlineplus.gov/ency/article/001577.htm
- Encyclopedia: Hydrocephalus
  https://medlineplus.gov/ency/article/001571.htm
- Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm
- Encyclopedia: Spinal Stenosis
  https://medlineplus.gov/ency/article/000441.htm
- Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center

- Achondroplasia
  https://rarediseases.info.nih.gov/diseases/8173/achondroplasia

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/a/achondroplasia
- Disease InfoSearch: Achondroplasia
  http://www.diseaseinfosearch.org/Achondroplasia/113

- Genetics Education Materials for School Success (GEMSS)
  http://www.gemssforschools.org/conditions/achondroplasia/default
- Johns Hopkins Medicine
  http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/achondroplasia.html
- KidsHealth from the Nemours Foundation
  http://kidshealth.org/en/parents/dwarfism.html
- MalaCards: achondroplasia
  http://www.malacards.org/card/achondroplasia
- Nemours Children's Health System
  http://www.nemours.org/service/medical/skeletal-dysplasia/achondroplasia.html?tab=about
- Orphanet: Achondroplasia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=15
- University of Virginia Health System
  https://uvahealth.com/services/endocrine-system/endocrine-conditions/achondroplasia

Patient Support and Advocacy Resources

- Human Growth Foundation
  http://hgfound.org/
- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Little People of America, Inc.
  http://www.lpaonline.org
- March of Dimes
  http://www.marchofdimes.org/baby/achondroplasia.aspx
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/achondroplasia/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dwarfism.html
- The MAGIC Foundation
  https://www.magicfoundation.org/

GeneReviews

- Achondroplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1152

Genetic Testing Registry

- Achondroplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001080/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22achondroplasia%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Achondroplasia%5BMAJR%5D%29+AND+%28achondroplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- ACHONDROPLASIA
  http://omim.org/entry/100800

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/achondroplasia

Reviewed: May 2012
Published: February 7, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services